



cyclic neutropenia

Cyclic neutropenia is a disorder that causes frequent infections and other health problems in affected individuals. People with this condition have recurrent episodes of neutropenia during which there is a shortage (deficiency) of neutrophils. Neutrophils are a type of white blood cell that plays a role in inflammation and in fighting infection. The episodes of neutropenia are apparent at birth or soon afterward. For most affected individuals, neutropenia recurs every 21 days and lasts about 3 to 5 days.

Neutropenia makes it more difficult for the body to fight off pathogens such as bacteria and viruses, so people with cyclic neutropenia typically develop recurrent infections of the sinuses, respiratory tract, and skin. Additionally, people with this condition often develop open sores (ulcers) in the mouth and colon, inflammation of the throat (pharyngitis) and gums (gingivitis), recurrent fever, or abdominal pain. People with cyclic neutropenia have these health problems only during episodes of neutropenia. At times when their neutrophil levels are normal, they are not at an increased risk of infection and inflammation.

Frequency

Cyclic neutropenia is a rare condition and is estimated to occur in 1 in 1 million individuals worldwide.

Genetic Changes

Mutations in the *ELANE* gene cause cyclic neutropenia. The *ELANE* gene provides instructions for making a protein called neutrophil elastase, which is found in neutrophils. When the body starts an immune response to fight an infection, neutrophils release neutrophil elastase. This protein then modifies the function of certain cells and proteins to help fight the infection.

ELANE gene mutations that cause cyclic neutropenia lead to an abnormal neutrophil elastase protein that seems to retain some of its function. However, neutrophils that produce abnormal neutrophil elastase protein appear to have a shorter lifespan than normal neutrophils. The shorter neutrophil lifespan is thought to be responsible for the cyclic nature of this condition. When the affected neutrophils die early, there is a period in which there is a shortage of neutrophils because it takes time for the body to replenish its supply.

Inheritance Pattern

Cyclic neutropenia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In most cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Other Names for This Condition

- cyclic hematopoiesis
- cyclic leucopenia
- periodic neutropenia

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Cyclical neutropenia
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0221023/>

Other Diagnosis and Management Resources

- GeneReview: ELANE-Related Neutropenia
<https://www.ncbi.nlm.nih.gov/books/NBK1533>
- Seattle Children's Hospital
<http://www.seattlechildrens.org/medical-conditions/heart-blood-conditions/neutropenia-symptoms/>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Neutropenia--Infants
<https://medlineplus.gov/ency/article/007230.htm>
- Health Topic: Blood Disorders
<https://medlineplus.gov/blooddisorders.html>

Genetic and Rare Diseases Information Center

- Cyclic neutropenia
<https://rarediseases.info.nih.gov/diseases/6229/cyclic-neutropenia>

Educational Resources

- Disease InfoSearch: Cyclic Hematopoiesis
<http://www.diseaseinfosearch.org/Cyclic+Hematopoiesis/2064>
- KidsHealth from Nemours
<http://kidshealth.org/en/parents/neutropenia.html>
- Lurie Children's Hospital of Chicago
<https://www.luriechildrens.org/en-us/care-services/conditions-treatments/neutropenia/Pages/index.aspx>
- Merck Manual for Health Care Professionals
<http://www.merckmanuals.com/home/blood-disorders/white-blood-cell-disorders/neutropenia>
- Monroe Carell Jr. Children's Hospital at Vanderbilt: All About the Immune System
<http://www.childrenshospital.vanderbilt.org/library/article.php?ContentTypeId=90&ContentId=P01665&Category=SearchTitle§ion=33153&term=i&>
- Orphanet: Cyclic neutropenia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2686

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/cyclic-neutropenia/>
- The Severe Chronic Neutropenia International Registry: Cyclic Neutropenia
<http://depts.washington.edu/registry/Info.Cyclic.Neut.htm>

GeneReviews

- ELANE-Related Neutropenia
<https://www.ncbi.nlm.nih.gov/books/NBK1533>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22cyclic+neutropenia%22+OR+%22Neutropenia%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Neutropenia%5BMAJR%5D%29+AND+%28cyclic+neutropenia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

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<http://omim.org/entry/162800>

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